



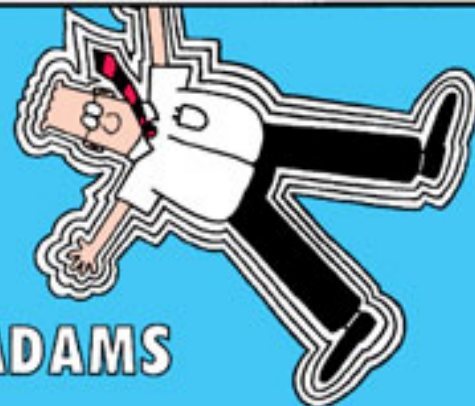
Rare Disease Conference Report

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BY
SCOTT ADAMS



Promoting **Quality**

Laboratory Testing for Rare Diseases: Keys to Ensuring Quality Genetic Testing



May 19-21, 2004
Atlanta, GA





Rare Disease Testing Conference: May 19-21, 2004

- Assure access to quality laboratory testing
 - Improve oversight of research laboratories
- Expedite translation of gene findings into clinical and public health practice
- Identify data and education needs
- Promote collaboration, cooperation, partnership, and community involvement



What is a rare disease or condition?

- NIH Office of Rare Diseases
 - 1 in every 12 individuals in US
- Orphan Drug Act:
 - $< 1 / 1,465$ (affect $< 200,000$ in US)
- Safe Medical Devices Act of 1990
 - $< 1 / 1,000 / \text{yr}$ ($< 4000 / \text{y}$ in US)
- Orphanet
 - < 1 per 2,000 people in Europe
- 5 new diseases every week in medical literature:
Over 6,000 diseases known today



Current Genetic Tests

GeneTests: April 2004

Total Tests

1,039

Clinical

694 (67%)

US: 542 (78%)

Non US Only: 152 (22%)

Research Only

354 (33%)



Testing Laboratories

GeneTests: April 2004

Total Laboratories

598

US

412 (69%)

Clinical

247 (60%)

Research Only

165 (40%)

Non US

186 (31%)

- “Research only” labs account for 40% of US labs listed in GeneTests
- Non US labs account for 31% of all labs listed in the directory



Testing Availability

GeneTests April 2004

Total Clinical Testing

694 Diseases

Testing available from only 1 lab

308 (44%)

Testing available from 2-5 labs

224 (32%)

Subtotal

532 (76%)

- CAP Molecular Genetics Survey: 17 tests
- EMQN: 13 tests, 1 sequencing



Summary

- Human genome: ~35,000 genes
- Genes with known sequence as of May 2004: 11,550
- New OMIM entries: 60-100 per month
- Current rare diseases: 6,000 – 7,000
- New rare diseases: ~20 per month (5/wk)
- Diseases for which clinical testing is available: 694
- New testing: <10 per month (2 in April 2004)



Rare Disease Conference Outcomes

- Formed North American Rare Disease Network
 - All network laboratories CLIA certified
 - Reports with limitations from CLIA laboratory
 - Work collectively to develop new tests
 - Foster research/clinical laboratory partnerships
 - Backup for sole source tests



Rare Disease Conference Outcomes

- American Society of Human Genetics and Office for Human Research Protections to provide education for researchers and IRBs
- NIH pilot programs to fund translation of research tests into clinically applicable tests
- 2005 meeting planned to assign responsibility for additional areas of focus – communication, coordination, roles
- Website: <http://www.phppo.cdc.gov/dls/genetics>

Pathways to Quality for Rare Disease Testing and Testing Materials

